Molecular Study in Myeloid Neoplasms (Atypical Chronic Myeloid Leukemia, Chronic Myelomonocytic Leukemia, Chronic Neutrophilic Leukemia, Myelodysplasia/Myeloproliferative Neoplasm): Multicenter Study (King Memorial Chulalongkorn Hospital and Sunpasitthiprasong Hospital)

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Background: Myeloid disorders are heterogeneous diseases. Diagnosis of these entities can be made using WHO 2016 criteria. However, making definite diagnosis can be uncertain and difficult in some cases. Recently, there have been studies of molecular abnormalities detected in myeloid disorders. CSF3R mutations are detected approximately 60% in chronic neutrophilic leukemia and atypical CML (aCML). Whereas, ETNK1 and SETBP1 mutations are detected about 8.8% and 24% in CML, respectively. SETBP1 mutations are also found 10% in MDS/MPN-U and 4% in CMML.

Objective: This study aimed to detect 3 novel gene mutations (CSF3R, ETNK1 and SETBP1) in 4 myeloid disorders (aCML, CNL, CMML and MDS/MPN-U) to help in making diagnosis.

Methods: We collected blood or bone marrow from patients with clinical and laboratory parameters compatible to WHO 2016 diagnostic criteria of CML, CNL, CMML, and MDS/MPN-U. DNA was extracted from the specimen and sequenced for CSF3R, ETNK1 and SETBP1 by direct sequencing technique to detect previously reported mutations.

Results: There were 14 patients with myeloid disorders. Five cases were diagnosed as CMML, followed by 2 cases of CNL, 1 case of CML, and 6 cases of MDS/MPN-U. Non-synonymous mutation of ETNK1 (c.A731G, p. N244S) was detected in 1 case of CMML. Recurrent mutation of SETBP1 (c.G2602A, p.D868N) was detected in 1 case of MDS/MPN-U, and 1 case of CNL. Non-synonymous mutation of CSF3R (T618I) was detected in 1 case of CNL. In total, we detected 3 mutations in 14 cases, accounting for 21%.

Conclusion: Prevalence of 3 gene mutations in CML, CNL, CMML and MDS/MPN-U is quite low (21% of all cases). However, somatic mutations can help support diagnosis in myeloid disorders.

Keywords: Somatic mutation, Myeloid disorders, Sequencing